B3GLCT gene

beta 3-glucosyltransferase

Normal Function

The *B3GLCT* gene (formerly known as *B3GALTL*) provides instructions for making an enzyme called beta 3-glucosyltransferase (B3Glc-T), which is involved in the complex process of adding sugar molecules to proteins (glycosylation). Glycosylation modifies proteins so they can perform a wider variety of functions. The B3Glc-T enzyme is involved in a two-step glycosylation pathway that results in the formation of a sugar structure, made up of the sugars fucose and glucose, on a specific location of several different proteins. The B3Glc-T enzyme is responsible for the second step, which adds a glucose molecule to the fucose molecule already attached to the protein. The *B3GLCT* gene is normally turned on (active) in most cells of the body, which suggests that the B3Glc-T enzyme plays an important role across many cell types.

Health Conditions Related to Genetic Changes

Peters plus syndrome

At least 10 mutations that cause Peters plus syndrome have been identified in the *B3GLCT* gene. Peters plus syndrome is characterized by eye abnormalities, short stature, intellectual disability, and distinctive facial features. The most common *B3GLCT* gene mutation replaces the DNA building block (nucleotide) guanine with the nucleotide adenine near an area of the gene called exon 8 (written as 660+1G>A). This mutation disrupts how genetic information is pieced together to produce the B3Glc-T enzyme. The resulting enzyme is abnormally short and nonfunctional. It is unclear how the loss of functional B3Glc-T enzyme leads to the signs and symptoms of Peters plus syndrome, but impaired glycosylation likely disrupts the function of many proteins, which may contribute to the variety of features.

Other Names for This Gene

- B3GALTL
- B3Glc-T
- B3GLT_HUMAN
- B3GTL
- beta 1,3-galactosyltransferase-like

- beta-3-glycosyltransferase-like
- beta3Glc-T

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of B3GLCT (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=145173[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28B3GALTL%5BTIAB%5D%29+OR+%28beta1,3-galactosyltransferase%5BTIAB%5D%29+OR+%28B3GLCT%5BTIAB%5D%29+OR+%28beta+3-galactosyltransferase%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- BETA-3-GLUCOSYLTRANSFERASE (https://omim.org/entry/610308)

Research Resources

- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=B3GLCT[gene])
- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/145173)

References

- Hess D, Keusch JJ, Oberstein SA, Hennekam RC, Hofsteenge J. Peters Plussyndrome is a new congenital disorder of glycosylation and involves defectiveOmicron-glycosylation of thrombospondin type 1 repeats. J Biol Chem. 2008 Mar21;283(12):7354-60. doi: 10.1074/jbc.M710251200. Epub 2008 Jan 16. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18199743)
- Kozma K, Keusch JJ, Hegemann B, Luther KB, Klein D, Hess D, Haltiwanger RS, Hofsteenge J. Identification and characterization of abeta1,3-glucosyltransferasethat synthesizes the Glc-beta1,3-Fuc disaccharide on thrombospondin type 1repeats. J Biol Chem. 2006 Dec 1;281(48):36742-51. Epub 2006 Oct 10. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17032646)
- Lesnik Oberstein SA, Kriek M, White SJ, Kalf ME, Szuhai K, den Dunnen JT, Breuning MH, Hennekam RC. Peters Plus syndrome is caused by mutations in B3GALTL,a putative glycosyltransferase. Am J Hum Genet. 2006 Sep;79(3):562-6. Epub 2006Jul 19. Erratum in: Am J Hum Genet. 2006 Nov;79(5):985. Citation on

PubMed (https://pubmed.ncbi.nlm.nih.gov/16909395) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1559553/)

- Lesnik Oberstein SAJ, Ruivenkamp CAL, Hennekam RC. Peters Plus Syndrome. 2007 Oct 8 [updated 2017 Aug 24]. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, BeanLJH, Gripp KW, Mirzaa GM, Amemiya A, editors. GeneReviews® [Internet]. Seattle(WA): University of Washington, Seattle; 1993-2022. Available fromhttp://www.ncbi.nlm.nih.gov/books/NBK1464/ Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/20301637)
- Reis LM, Tyler RC, Abdul-Rahman O, Trapane P, Wallerstein R, Broome D, HoffmanJ, Khan A, Paradiso C, Ron N, Bergner A, Semina EV. Mutation analysis of B3GALTL in Peters Plus syndrome. Am J Med Genet A. 2008 Oct 15;146A(20):2603-10. doi:10.1002/ajmg.a.32498. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18798333) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2755183/)
- Sato T, Sato M, Kiyohara K, Sogabe M, Shikanai T, Kikuchi N, Togayachi A,Ishida H, Ito H, Kameyama A, Gotoh M, Narimatsu H. Molecular cloning andcharacterization of a novel human beta1,3-glucosyltransferase, which is localizedat the endoplasmic reticulum and glucosylates O-linked fucosylglycan onthrombospondin type 1 repeat domain. Glycobiology. 2006 Dec;16(12):1194-206. Epub2006 Aug 9. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16899492)

Genomic Location

The *B3GLCT* gene is found on chromosome 13 (https://medlineplus.gov/genetics/chromosome/13/).

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